

EMMA Strain Original Submission Form

The original data have been submitted to INFRAFRONTIER on 2023-12-11 17:03:14.0

Bex3-Delta(24-72) / EM:14601

Genotype (step 1)

Strain name	Bex3-Delta(24-72)
Genetic description	CRISPR/Cas9 technology was used to target the mouse Bex3 gene located at chromosome X. Bex3-Delta(24-72) mice carry a 147-bp deletion in exon 3, which is the first and only coding exon. This deletion removes 49 amino acids of the central core of the protein, specifically the pro-apoptotic domain. The protein still retains the C-terminal coiled coil domain required for BEX3 dimerization, nuclear import, ubiquitination, and interaction with its multiple partners.
Current genetic background	CBA x C57BL/6
Breeding history	Line is maintained by sibling breeding with occasional outbreeding to the founder stock (CBA/C57Bl6 F1 hybrid)

Mutations

Type	EM
Affected gene/allele	Bex3
Affected allele	Bex3
Original genetic background	(C57BL/6 x CBA)F1

ONE mutant strain is defined by its specific set of mutation(s) and its specific genetic background. Therefore strains with the same set of mutation(s) but different backgrounds do require DISTINCT submission forms (i.e) ONE form for each background

Phenotype (step 2)

Phenotypic description of mice homozygous for the mutation	Bex3-Delta(24-72) homozygous mice display skull and brain abnormalities, as well as a plethora of behavioural and cognitive alterations. Of note, these phenotypes are less severe than in Bex3KO mice. At the molecular level, they present aberrant mTOR signalling in the brain. These phenotypes are reminiscent of human neurological disorders such as ASD or schizophrenia.
Phenotypic description of mice heterozygous or hemizygous for the mutation	Since Bex3 is located in the X chromosome, male hemizygous mutant mice display the same phenotype as homozygous females, which is described in the previous section. Heterozygous females show no apparent phenotype

Breeding and Animal welfare (step 3)

Are homozygous mice viable?	Yes
Are homozygous mice fertile?	Yes
Are heterozygous/hemizygous mice fertile?	Yes
Are homozygous matings required?	No
Breeding and reproduction details	Often, matings between homozygous mutant animals fail, presumably due to behavioural alterations. Thus, matings using heterozygous females -instead of homozygous mutants- are recommended.
Husbandry requirements	Due to behavioural alterations reminiscent of ASD, among other neurological disorders, environmental enrichment is recommended.
Are mice immunocompromised?	No
Animal welfare	

[Health report](#) (_health_2020A000000965_P1-6817.pdf)

Please note that under certain circumstances (e.g.: long-term cryopreservation by sperm freezing) the strain's original genotype will not always be available for future reconstitution of live colonies. Therefore, the original genetic background cannot be guaranteed.

Characterization (step 4)

[Genotyping file](#) (_pcr_Genotyping_protocol_Bex3-Delta(24-72).pdf)

Characterization by other method	
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References (step 5)

Has this mouse mutant strain been published or accepted for publication? Yes

PubMed ID	33100228
Description	Generation of the mutant mouse strain and description of the phenotype
Title	Characterization of an eutherian gene cluster generated after transposon domestication identifies Bex3 as relevant for advanced neurological functions.
Authors	Navas-Pérez E, Vicente-García C, Mirra S, Burguera D, Fernández-Castillo N, Ferrán JL, López-Mayorga M, Alaiz-Noya M, Suárez-Pereira I, Antón-Galindo E, Ulloa F, Herrera-Úbeda C, Cuscó P, Falcón-Moya R, Rodríguez-Moreno A, D'Aniello S, Cormand B, Marfany G, Soriano E, Carrión AM, Carvajal JJ, Garcia-Fernández J.
Journal/Book	Genome Biol
Year	2020
Volume	21
Pages	267
Other description	
No PubMed details	

Research value (step 6)

Does this strain model a human condition or disease?	Yes
Additional information	

Disease ontology

- schizophrenia / DOID:5419
- X-linked disease / DOID:0050735
- autism spectrum disorder / DOID:0060041

Research areas

- Models, Neurological

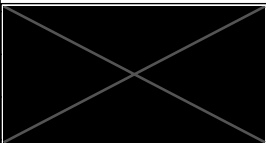
Research tools

Additional information (step 7)

MTA File uploaded	
Are there any intellectual property rights or patented technologies linked to this strain?	Yes
IPR details?	CRISPR/Cas9 technology was used to produce this line
Is the producer the exclusive owner of this strain (including all original materials, e.g. alleles or constructs, used for its generation)?	Yes
Do you have permission from all owners to deposit this strain in the EMMA repository?	Yes
Producer ILAR code	
Is this strain being deposited with any other institution or company intending to make it available for distribution?	No
Are other laboratories producing similar strains?	Not known
How many requests for this strain have you received in the last 6 months	0
Do you require delayed release for your strain (max. two years grace period)?	No
How many mice of breeding age could you provide (Male/Female)	6/0
When can you provide the mice of breeding mice?	11/2021

Additional materials of interest

Submitter information (step 8)

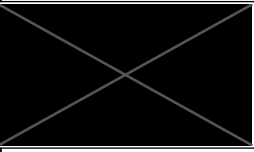
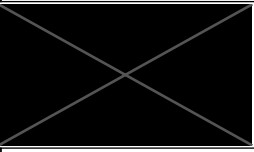
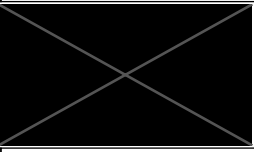
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The [EMMA repository conditions](#) and [data privacy statement](#) have been read, understood and agreed to.